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News Release

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Combining genetic and clinical factors may better identify those at risk for atherosclerotic cardiovascular disease

BOSTON – Combining genetic and clinical factors may better identify those at risk for atherosclerotic cardiovascular disease, according to researchers at VA Boston Healthcare System in a study published online May 3, 2023, in JAMA Cardiology.

“Currently, clinicians use risk calculators to estimate a patient’s risk for atherosclerotic cardiovascular disease based on risk factors identified during clinic visits, but these prediction models can overestimate or underestimate a patient's risk level, resulting in over- or under-treatment of risk factors such as cholesterol levels,” said Dr. Jason Vassy co-first author of the study and attending physician at VA Boston HCS.

Atherosclerotic cardiovascular disease – or ASCVD – which includes myocardial infarction, ischemic stroke and deaths related to coronary heart disease, is a major source of illness and death worldwide. The researchers wanted to see if adding polygenic risk scores – or PRS – which are numerical scores based on genetic variants associated with a disease, to a traditional clinical risk prediction model could improve prediction of ASCVD risk.

Participants were 79,151 Veterans enrolled in VA’s Million Veteran Program, known as MVP, who volunteered to share their genetic and medical record data for research. Using participant data, the researchers built risk models to predict which Veterans would have an ASCVD event. Researchers then compared the traditional clinical risk prediction model to one that also included the PRS.

Researchers reviewed participant data from their MVP enrollment in 2011 through Dec. 31, 2018. During this timeframe, 5,485 Veterans developed ASCVD. The risk prediction model that combined genetics and traditional clinical risk factors performed slightly better overall across all three ancestry groups – Black, Hispanic and white – compared with the traditional clinical model. The inclusion of genetics in the risk prediction model improved risk prediction for women and younger ages, suggesting that clinical value for women and younger populations may be higher.

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Combining PRS and clinical factors to identify ASCVD risk – 2 of 2

“These results are encouraging, and we hope incorporating polygenic risk scores into prediction models will someday help identify those most at risk for atherosclerotic cardiovascular disease, so we can better implement prevention strategies, like taking statin medications,” said Dr. Daniel Posner, co-first author, biostatistician and researcher at VA Boston HCS. “More research is needed, however, before definitive conclusions can be drawn, particularly clinical trials.”

The study is available at <https://jamanetwork.com/journals/jamacardiology/article-abstract/2804439>